Treacher Collins syndrome: A case study

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Treacher Collins syndrome is a disorder of craniofacial development with high penetrance and variable expressivity. Its incidence is approximately 1 in 50,000 live births. In this article, we describe the orthodontic treatment of an 11-year-old boy with Treacher Collins syndrome. (Am J Orthod Dentofacial Orthop 2014;146:665-72)

Treacher Collins syndrome (TCS), also known as mandibulofacial dysostosis and Franceschetti-Zwahlen-Klein syndrome, is a disorder of craniofacial development with high penetrance and variable expressivity.[1,2] Mildly affected persons might be diagnosed only retrospectively, after the birth of a more severely affected family member. In contrast, severe cases may lead to perinatal death.[3] Airway obstruction and feeding difficulties result from severe mandibular hypoplasia and the resulting glossoptosis in more severely affected neonates. The incidence of TCS is approximately 1 in 50,000 live births. Generally, it is characterized by symmetrical developmental anomalies of tissues derived from the first and second embryonic branchial arches.[4]

TCS occurs as a result of a mutation in 1 of 3 genes: *TCOF1*, *POLR1C*, or *POLR1D*.[5] Mutation of the *TCOF1* gene is most commonly found, accounting for 78% to 93% of TCS cases. Mutations of *POLR1C* and *POLR1D* account for approximately 8%. TCS caused by heterozygous mutation in *TCOF1*, or less commonly *POLR1D*, is inherited in an autosomal dominant manner, whereas compound heterozygous mutations in *POLR1C* are inherited in an autosomal recessive manner. Autosomal dominant inheritance accounts for the majority of TCS cases.[5] It is estimated that 60% of autosomal dominant cases of TCS arise as the result of de novo mutations, and 40% are familial.[1]

The severity of the phenotype for TCS is highly variable. Extremely common craniofacial features of TCS include zygomatic hypoplasia (81%) and micrognathia (78%). In severe cases, the zygomatic arches may be absent.[3] Alterations in the size, shape, and position of the external ears are frequently associated with atresia of the external auditory canals. Anomalies of the middle ear ossicles often result in conductive hearing loss, whereas mixed or sensorineural hearing loss is rare in TCS patients.[3] Affected patients may have a Tessier number 6 facial cleft extending from the orbital rim to the zygoma. Antimongoloid slant of the palpebral fissures, lower eyelid coloboma, and partial absence of eyelashes are also commonly observed.[6]

There are multiple oral manifestations of TCS. Examples include high palatal vault, clefting of the palate, and enamel hypoplasia. Skeletal hypoplasia of the maxilla and mandible often leads to an anterior open bite. Dental malocclusion involving malpositioned and reduced number of teeth is frequently observed. Tooth agenesis, mainly mandibular second premolars, and ectopic eruption of the maxillary first molars are also common. [6]

DIAGNOSIS AND ETIOLOGY

An 11-year-old boy first came to the Division of Orthodontics at Columbia University College of Dental Medicine in New York City with a chief complaint of "crooked teeth." He was referred for orthodontic treatment after an evaluation by the craniofacial team at the medical center. He had been receiving craniofacial care at New York-Presbyterian Morgan Stanley Children's Hospital at Columbia University Medical Center. His medical history was positive for TCS, although his 2 siblings were not affected by the syndrome. Multiple ear infections and snoring were also documented. The surgical history included a temporary tracheostomy

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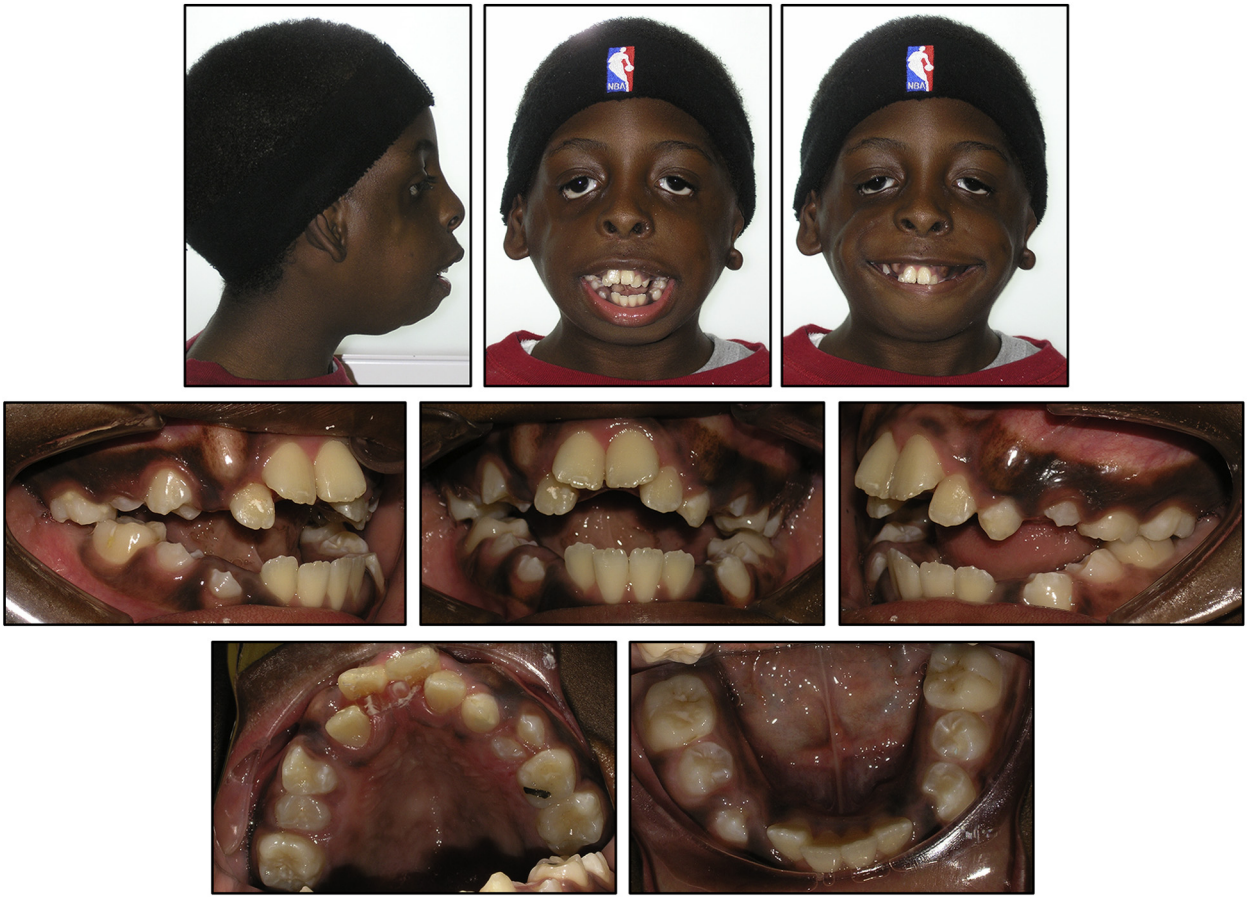

Fig 1. Pretreatment photographs.

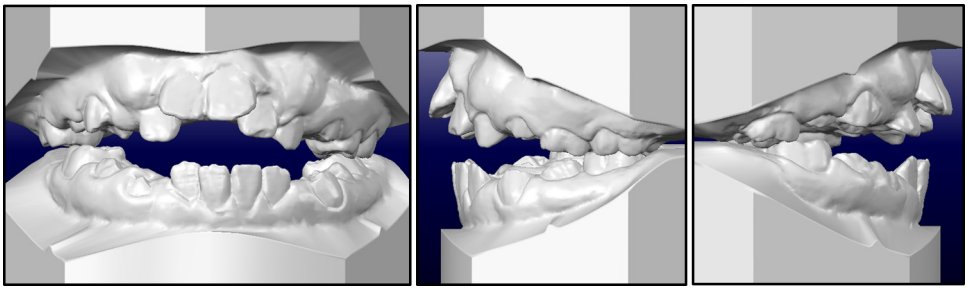

Fig 2. Pretreatment digital dental cast photographs.

during infancy, right myringotomy and tube placement, and multistage left ear reconstructions.

The extraoral examination showed left microtia, malar hypoplasia, micrognathia, convex profile, and severe lip incompetence. The intraoral examination showed an anterior open bite of 10 mm, occlusion only on the first molars, right side Class III molar relationship, left side Class I molar relationship, maxillary transverse discrepancy, and significant arch length-tooth size discrepancy (Figs 1 and 2). He also had a limited range of motion in performing lateral and protrusive movements.

Panoramic (Fig 3) and lateral cephalometric (Fig 4) radiographs were obtained, and a cephalometric analysis was performed using the Columbia analysis.[7] This analysis (Table) showed a low SNB angle, which supports the micrognathia observed clinically, and a high ANB angle. The steep mandibular plane angle, represented by the

Table. Columbia analysis of the cephalometric radiographs

	Pretreatment				*Posttreatment*			
Measurement	*Value*	*Norm*	*SD*	*Deviation from norm*	*Value*	*Norm*	*SD*	*Deviation from norm*
SNA (°)	80.8	81.0	4.0	−0.0	78.1	82.0	4.0	−1.0
SNB (°)	64.7	78.0	3.0	−4.4	65.8	80.0	3.0	−4.7
ANB (°)	16.2	3.0	2.5	5.3	12.3	2.0	2.5	4.1
Wits appraisal (mm)	8.1	1.0	3.0	2.4	3.0	1.0	3.0	0.7
SN-GoGn (°)	65.2	32.0	5.0	6.6	55.5	32.0	5.0	4.7
SN-palatal (SN-PP) (°)	16.3	8.2	3.3	2.5	16.3	8.2	3.3	2.5
Palatal-mand angle (PP-GoGn) (°)	48.9	22.0	6.0	4.5	39.1	23.0	6.0	2.7
Y-axis (SGn-SN) (°)	91.7	67.0	5.5	4.5	87.8	67.0	5.5	3.8
P-A face height (S-Go/N-Me) (%)	51.7	65.0	4.0	−3.3	50.1	65.0	4.0	−3.7
UAFH/LAFH ratio (N-ANS/ANS-Me) (%)	64.3	80.0	7.0	−2.2	68.1	80.0	7.0	−1.7
U1-SN (°)	99.2	103.5	5.0	−0.9	83.2	105.0	5.0	−4.4
Interincisal angle (U1-L1) (°)	95.1	130.0	5.0	−7.0	123.5	130.0	5.0	−1.3
L1-GoGn (°)	100.4	93.0	6.0	1.2	97.8	93.0	6.0	0.8
L1 protrusion (L1-APo) (mm)	16.7	1.0	2.0	7.9	10.7	0.0	2.0	5.3
L1-NB (mm)	23.3	4.0	1.8	10.7	18.4	4.0	1.8	8.0
Pog-NB (mm)	−4.0	4.0	1.5	−5.3	0.9	4.0	1.5	−2.1
Holdaway angle (NB to H-line) (°)	23.7	8.0	4.0	3.9	23.5	8.0	4.0	3.9
Holdaway ratio (L1-NB:Pg-NB) (%)	−5.8	1.0	1.0	−6.8	20.3	1.0	1.0	19.3

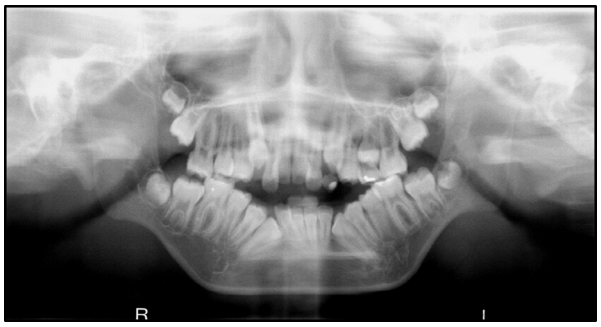

Fig 3. Panoramic radiograph.

SN-GoGn value of 65.2°, was greater than 6 SD from the norm of 32° (±5°). This was consistent with the syndrome.

TREATMENT OBJECTIVES

The treatment objectives for this patient required both orthodontic and surgical treatment, including correction of the maxillary transverse discrepancy, elimination of dental crowding, correction of the open bite, establishment of overbite and overjet for proper function, and improvement of the soft-tissue profile.

TREATMENT ALTERNATIVES

Expansion of the maxillary arch to correct the transverse discrepancy, and extraction of permanent teeth to correct the arch length tooth-size discrepancy were necessary. Surgical treatment was needed to correct the open bite and improve the soft-tissue profile. Mandibular distraction osteogenesis was considered to address the micrognathia. However, because the mandibular plane angle was extremely steep and a significant counterclockwise rotation of the mandibular body was desired to decrease the mandibular plane angle, bilateral extraoral inverted "L" ramus osteotomies were performed for the mandible.

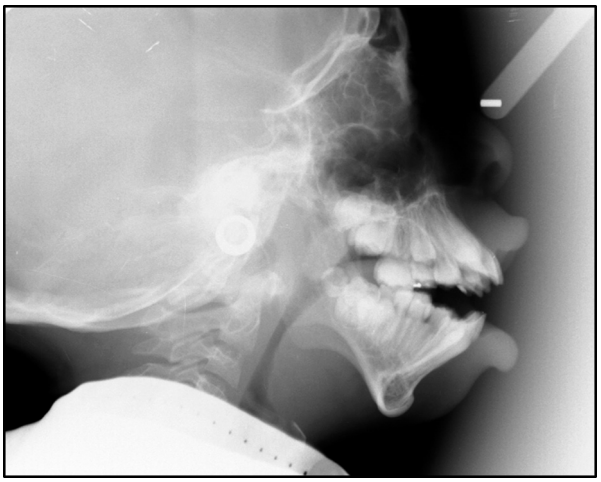

Fig 4. Lateral cephalometric radiograph.

TREATMENT PROGRESS

Presurgical orthodontic treatment was initiated by rapid maxillary expansion. A fan-type rapid maxillary

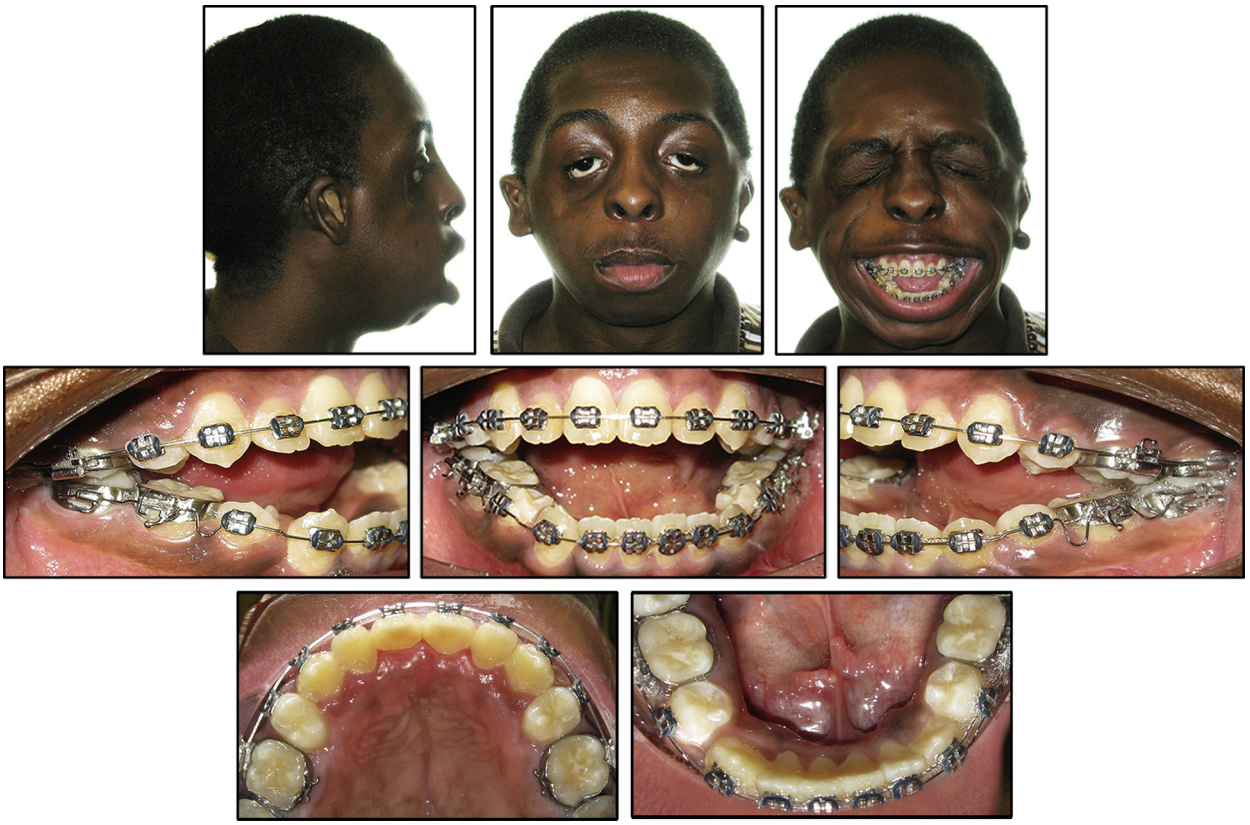

Fig 5. Presurgical photographs.

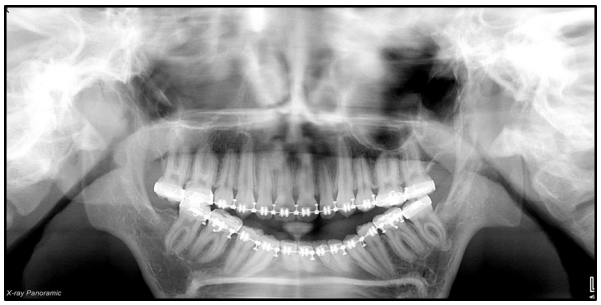

Fig 6. Presurgical panoramic radiograph.

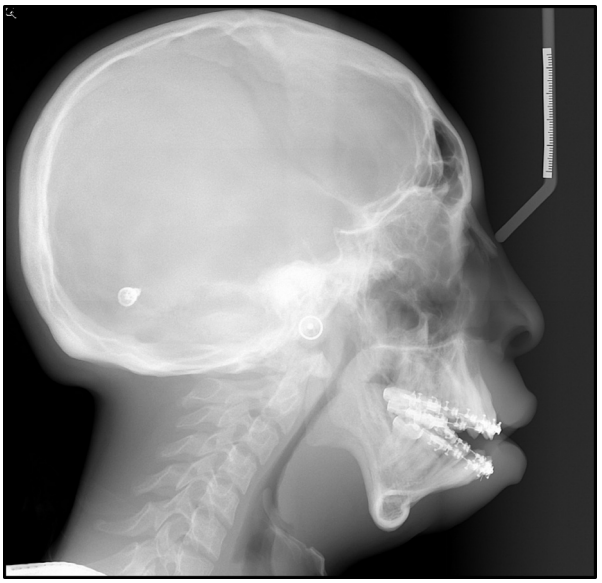

Fig 7. Presurgical lateral cephalometric radiograph.

expander, commonly the treatment of choice when greater intercanine width expansion is desired compared with intermolar width expansion, was used.[8] After expansion was achieved, both arches were bonded with standard edgewise appliances. The first premolar in each quadrant was extracted, creating space for the impacted canines. The mandibular canines were uprighted and brought into the dental arch. Presurgical orthodontic treatment continued until the patient underwent orthognathic surgery at 18 years of age. Figures 5-7 show the presurgical composite photographs, panoramic radiograph, and lateral cephalometric radiograph, respectively. The surgery was planned (Figs 8 and 9) with the Virtual Surgical

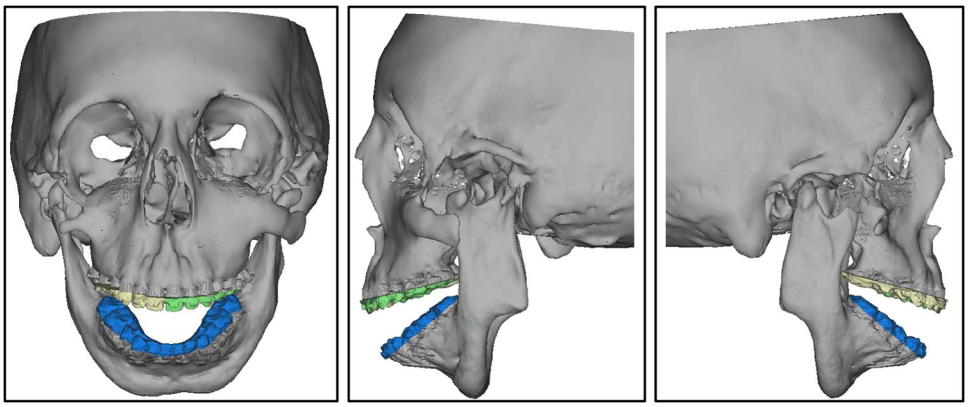

Fig 8. Surgery was planned with the Virtual Surgical Planning system and computer-assisted surgical simulation.

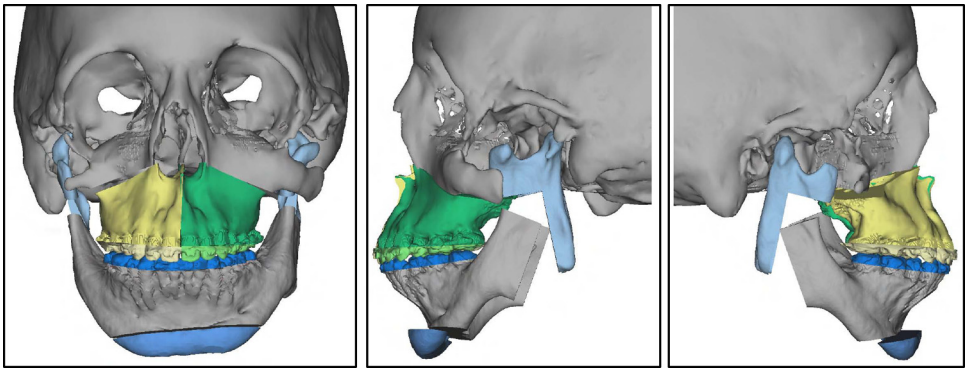

Fig 9. Final surgical treatment plan prediction.

Planning system (Medical Modeling, Golden, Colo) and computer-assisted surgical simulation as described by Hsu et al.[9] Surgical guides to design the ramus bone grafts, as well as the intermediate and final splints, were manufactured using the Virtual Surgical Planning system.

After orthodontic preparation, the patient underwent the following procedures: (1) temporary tracheostomy due to anticipated difficulty with intubation, (2) bilateral extraoral inverted "L" ramus osteotomies with a bone graft harvested from the left iliac crest, (3) LeFort 1 osteotomy (2 piece), and (4) genioplasty. Rigid fixation was used to retain the surgical movements. Figures 10 and 11 show the postsurgical composite photographs and lateral cephalometric radiograph, respectively.

Postsurgically, the patient continued with the finishing stages of orthodontic treatment in light round wires. Upon debonding of the fixed appliances, he was given a tooth positioner for retention. Tooth positioners can serve as the final means of retention and provide effective short-term adjunctive therapy for enhancing the finish. According to Park et al,[10] positioners increase the quality of the final occlusion. The improvement is not primarily due to an increase in occlusal contacts but instead is achieved by improving the first-order alignment. Final records were taken after debonding of all fixed appliances (Figs 12-14), and the lateral cephalogram was analyzed (Table).

TREATMENT RESULTS

Upon completion of orthodontic treatment with fixed appliances, correction in the transverse, anteroposterior, and vertical dimensions was achieved. The maxillary arch was expanded to achieve an ideal buccal overjet of the posterior teeth. The anterior open bite was addressed both orthodontically and surgically, and treatment was completed with positive overbite and overjet. Crowding was eliminated by extraction of the first premolars, and proper alignment of the remaining teeth was achieved. The soft-tissue

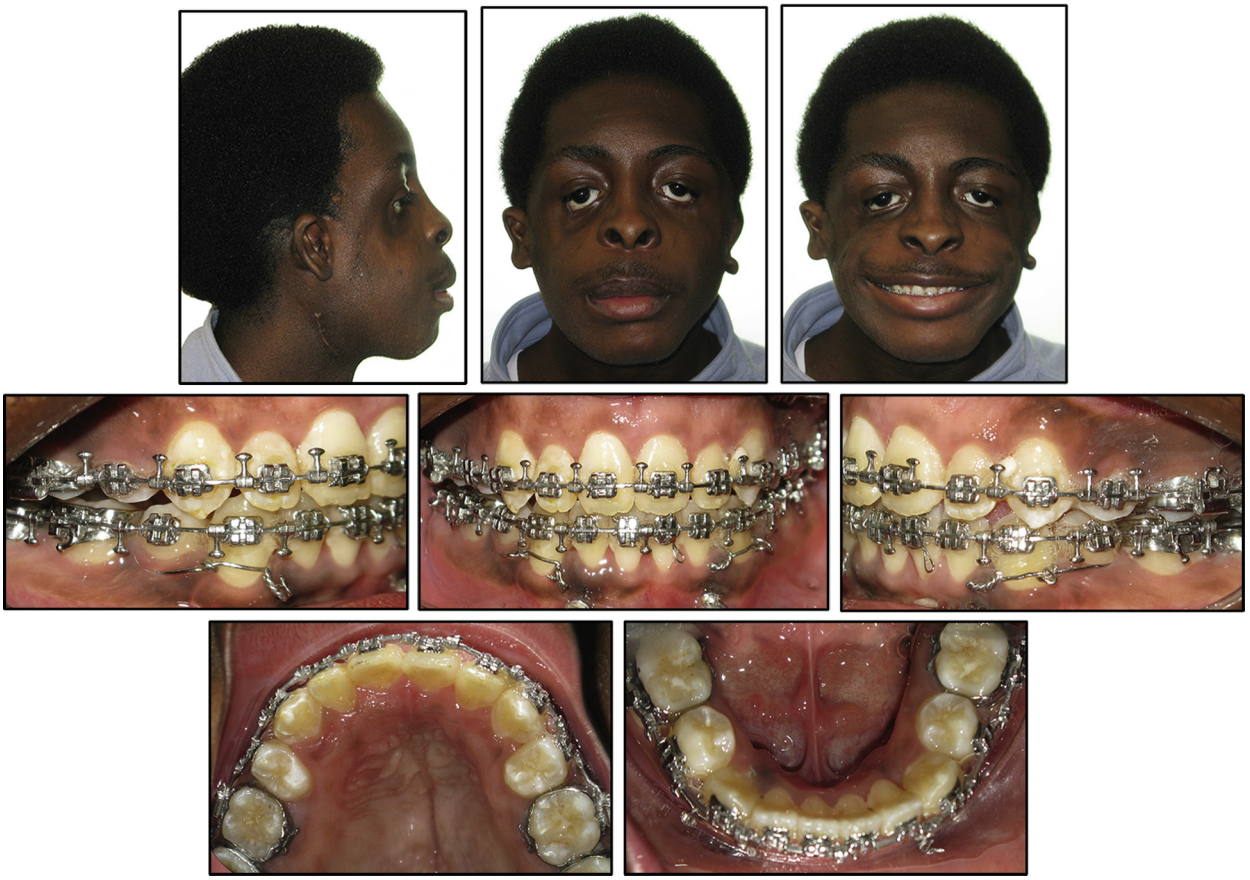

Fig 10. Postsurgical composite photographs.

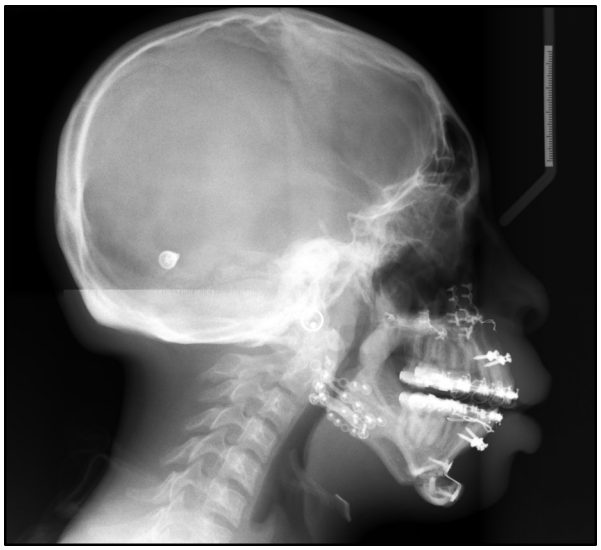

Fig 11. Postsurgical lateral cephalometric radiograph.

profile was improved surgically as the mandibular plane was rotated counterclockwise and chin projection was enhanced. Most importantly, the patient expressed great satisfaction with his smile and improved occlusion.

DISCUSSION

TCS is a congenital disorder of craniofacial development. Because it demonstrates high penetrance and extreme variation in expressivity of phenotype, some TCS patients may possess only mild characteristics, whereas others can be affected with severe deformations. Our patient demonstrated many of the clinical features described, including zygomatic hypoplasia, micrognathia, antimongoloid slant of palpebral fissures, atresia of the external auditory canal, and severe dental malocclusion.

Comprehensive care for TCS patients involves a team of multidisciplinary health care professionals

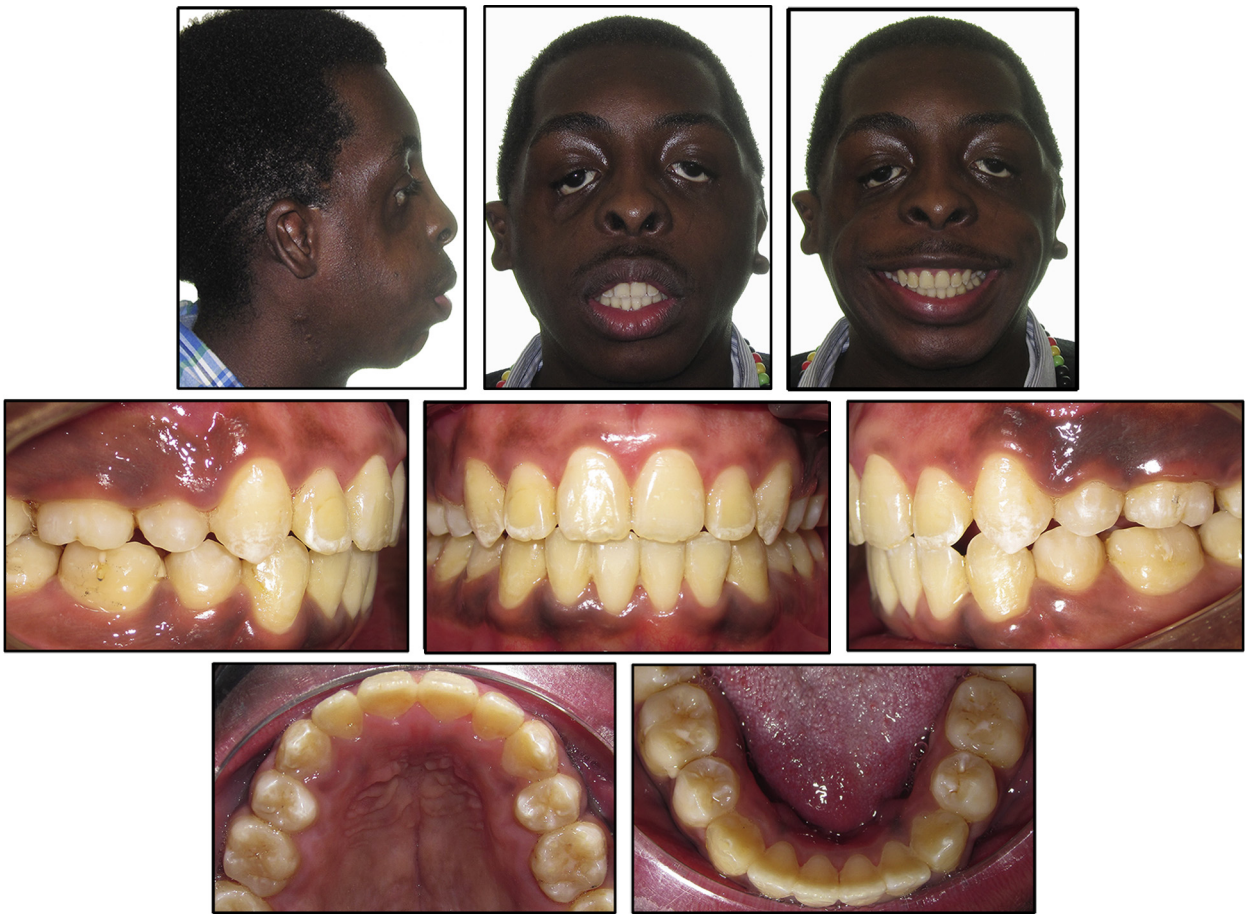

Fig 12. Posttreatment photographs.

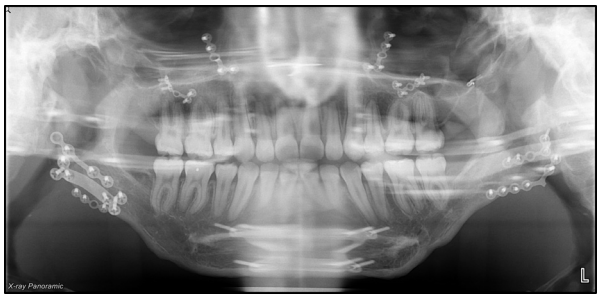

Fig 13. Posttreatment panoramic radiograph.

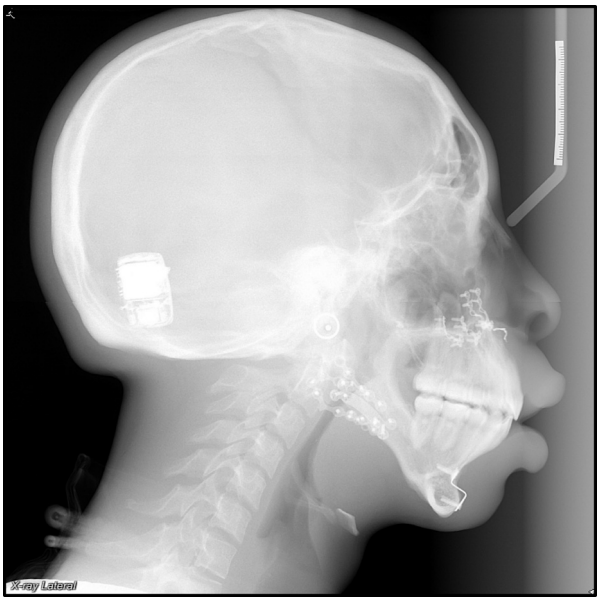

Fig 14. Posttreatment lateral cephalometric radiograph.

including craniofacial surgeons, pediatric dentists, orthodontists, otolaryngologists, ophthalmologists, speech pathologists, and psychiatrists. A primary concern regarding patients affected by TCS is a compromised upper airway. Micrognathia and tongue obstruction of the hypopharynx can cause respiratory difficulty from birth, and emergency surgery in the form of tracheostomy

or distraction osteogenesis might be necessary. Subsequently, multiple surgeries are usually needed to manage the hard and soft tissues involved.[3] Hearing should be tested before the age of 1 year because it is critical for speech development. Hearing loss and speech problems pose obstacles for developing learning ability, self-esteem, and social interaction and therefore must be treated.[3] Another concern for patients with TCS is obstructive sleep apnea, which has been documented in 25% of patients with TCS. Congenital velopharyngeal incompetence has also been documented in 30% to 40% of patients.[2] Our patient required a tracheostomy at birth and again at surgery. The postoperative lateral cephalogram demonstrated significant improvement in his airway; this was consistent with the resolution of his obstructive sleep apnea.

CONCLUSIONS

Patients with TCS, especially those severely affected, have complex functional disabilities and esthetic challenges. Early diagnosis and coordinated care by a craniofacial team help to ensure optimal results. Recent technological advances in treatment planning and manufacturing of computer-generated surgical guides were critical in the successful management of this patient.

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